

Health Care Provider Fact Sheet

Disease Name

Carnitine transporter deficiency

Alternate name(s)

Systemic carnitine deficiency, Carnitine deficiency, Carnitine uptake deficiency

Acronym

SCD, CUD

Disease Classification

Fatty Acid Oxidation Disorder

Variants

N/A

Variant name

N/A

Symptom onset

Infancy or childhood with fasting hypoglycemia, weakness and/or cardiomyopathy.

Symptoms

Hypoketotic hypoglycemia, seizures, vomiting, lethargy progressing to coma. Chronic muscle weakness, cardiomyopathy, hepatomegaly.

Natural history without treatment

Non-progressive developmental delay due to hypoglycemia, cardiomyopathy and muscle weakness.

Natural history with treatment

Developmental delay, if present, is not reversed by treatment. Cardiomyopathy and muscle weakness can be reversed by treatment.

Treatment

Carnitine supplementation, no fasting.

Other

N/A

Physical phenotype

Cardiomyopathy, muscle weakness.

Inheritance

Autosomal recessive

General population incidence

1/40,000

Ethnic differences

No

Population

N/A

Ethnic incidence

N/A

Enzyme location

Muscle, heart, kidney, leukocytes and fibroblasts

Enzyme Function

Transports carnitine into cells

Missing Enzyme

Carnitine transporter

Metabolite changes

Decreased free carnitine in plasma, increased carnitine in urine, decreased carnitine in muscle.

Gene

SLC22A5

Gene location

5q33.1

DNA testing available

May be available on a research basis.

DNA testing detail

If a mutation in a proband is detected, DNA carrier screening is possible. Protein analysis in cultured amniocytes, biochemical analyte testing. If a mutation in a proband is detected, DNA prenatal diagnosis via CVS or amniocytes is possible.

Prenatal testing

MS/MS Profile

Reduced concentrations of free carnitine and various acylcarnitine species.

OMIM Link

<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=212140>

Genetests Link

www.genetests.org

Support Group

FOD Family Support Group

<http://www.fodsupport.org>

Save Babies through Screening Foundation

<http://www.savebabies.org>

Genetic Alliance

<http://www.geneticalliance.org>